

CLAIMS

1. A diagnostic test kit for detecting the presence of or predisposition for breast cancer, whereby a means is provided for detecting a deletion of a stretch of nucleotides from a BRCA 1 gene in a sample.
- 5 2. A diagnostic test kit according to claim 1 whereby the means comprises at least one probe for hybridization.
3. A diagnostic test kit according to claim 2 whereby the means comprise the necessary elements for Southern blotting.
- 10 4. A diagnostic test kit according to claim 2 or 3 whereby the probe comprises a sequence complementary to sequences on both sides of the deletion in the BRCA 1 gene.
5. A diagnostic test kit according to anyone of the foregoing claims whereby the deletion comprises one or more
- 15 exons of the BRCA1 gene.
6. A diagnostic test kit according to anyone of the foregoing claims whereby the deletion comprises a frameshift and/or a termination codon.
7. A diagnostic test kit according to anyone of the
- 20 foregoing claims whereby the deletion comprises at least a major part of exon 22.
8. A diagnostic test kit according to anyone of the foregoing claims whereby the deletion comprises a major part of nucleotides 1396-1662.
- 25 9. A diagnostic test kit according to anyone of the foregoing claims whereby the deletion comprises at least a major part of exons 13-16.
10. A diagnostic test kit according to anyone of the foregoing claims whereby the deletion comprises at least a
- 30 major part of exon 13.
11. A diagnostic test kit according to anyone of the foregoing claims whereby the deletion comprises a deletion of a stretch of nucleotides between two ALU-elements.

12. A probe for use in a diagnostic test kit according to anyone of the foregoing claims comprising a nucleic acid sequence which is a fusion of two ALU elements of the BRCA1 gene.

5 13. A probe for use in a diagnostic test kit according to anyone of claims 1-11, which is a fusion product of two sequences adjacent to the site of a deletion of a stretch of nucleotides.

14. A method for determining the presence in a sample of  
10 a nucleic acid derived from a BRCA1 gene having a deletion of  
a stretch of nucleotides, comprising contacting said sample  
with at least one probe which alone or together with other  
means is capable of distinguishing between BRCA1 genes having  
said deletion and BRCA1 genes not having said deletion,  
15 allowing for possible hybridization between said probe and  
said nucleic acid and identifying the hybridization product.

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